

## What is PHACE syndrome?

PHACE syndrome is a group of disorders. It is identified by a large infantile hemangioma of the face, scalp, and neck. There is also a combination of developmental defects of the brain, blood vessels, eyes, heart, and/or chest wall to. The term PHACE is a word formed from the first letters of the words in the name (acronym).

## What does PHACE stand for and what are the possible problems?

- P =** Posterior fossa. In PHACE syndrome the structures at the back or hind part of the brain can be affected. Symptoms will vary depending on where the anomaly occurs. About half of patients with PHACE will have this problem.
- H =** Hemangioma. A hemangioma is a blood vessel birthmark. In PHACE syndrome the hemangioma often takes up a large area of the face or scalp. Almost all patients with PHACE will have a large hemangioma.
- A =** Abnormal head and neck arteries. Arteries are blood vessels that carry blood away from the heart to all parts of the body and brain. In PHACE syndrome there are abnormal shapes, sizes or paths the blood will travel through the neck and head. Four out of 5 children with PHACE have abnormal head or neck arteries.
- C =** Cardiac (heart) problems. These are called congenital heart defects. Congenital means they have been there since birth. In PHACE syndrome the large artery that comes directly out of the heart, called the aorta, may be narrow.
- E =** Eye problems. These are rare. The eyes may be small or have an unusual shape. There could be vision problems.



## What causes it?

The cause of PHACE syndrome is unknown. Research efforts are ongoing to find valuable information about PHACE syndrome so that care for these children can be improved.

## How is PHACE syndrome diagnosed?

There is no one sign, symptom or test that means your child has PHACE syndrome. The doctors will ask some questions and do an exam. Your child may need special Imaging (Radiology) tests. Tests that may be done are:

- Eye exam. An eye doctor will check vision and eye health.
- Echocardiogram (echo). This test uses ultrasound to look at the heart while it beats.
- MRI and MRA. Imaging is helpful to see possible problems in the brain, neck, chest, heart and eyes.

## Where can I get more information?

You can learn more at: <https://childrenswi.org/medical-care/birthmarks-and-vascular-anomalies-center/conditions/phace-syndrome>

Your child's healthcare team is always the best source for information about your child's conditions. It will be helpful for you to write your questions down before and after your visits. If your child is diagnosed with PHACE syndrome, it is important to see a multidisciplinary team of doctors that specialize with children with PHACE syndrome.

**ALERT:** Call your child's doctor, nurse, or clinic if you have any concerns or if your child:

- Is not eating like normal.
- Has to stop and rest often during a feeding.
- Has a blue color around the lips while eating.
- Is not gaining weight.
- Has a hemangioma that is not being treated and gets thicker.
- Has constant noisy breathing.
- Has a seizure.
- Has special health care needs not covered by this information.

**This sheet was created to help you care for your child or family member. It does not take the place of medical care. Talk with your healthcare provider for diagnosis, treatment and follow-up.**